

Table S1. Regulatory effects of the SNPs selected for the study and their effect on gene expression

SNPs	Regulatory effects according to HaploReg, SNPinfo, Regulome DB, VannoPortal										Expression QTLs (GTE _x) genes/ tissues	Splicing QTLs (GTE _x) genes/ tissues
	Prom histone marks	Enh histone marks	DNase	Altered reg motifs	TF Binding evidence	Rank / Score RegulomeDB	Splicing ESE/ ESS	SIFT/ CADD	Reg Score Vanno Portal	Reg Potential SNPinfo		
rs10490924 <i>ARMS2</i> missense	-	-	yes	15	5	5/0,13	–	Damaging	0.192	–	ARMS2: 8 PLEKHA1: 6 HTRA1: 1	ARMS2: 1 PLEKHA1: 1
rs2230199 <i>C3</i> missense	yes	yes	yes	28	29	3a/0,86	yes	Tolerated	0.584	0,33	GPR108: 7 CTD-3128G10.7: 1	-
rs2285714 <i>CFI</i> / <i>PLA2G12A</i> downstream variant	-	-	-	10	7	4/0,60	yes	Likely pathogenic	0.327	0,36	CFI: 1 PLA2G12A:23 MCUB:1 CASP6: 4	CFI: 33 PLA2G12A: 7
rs6677604 <i>CFH</i> intron variant	-	yes	yes	20	16	5/0,13	–	–	0.315	–	CFHR3: 27 CFHR1: 18 CFHR4: 1 ZBTB41: 1	CFH: 12 CFHR1: 3
rs800292 <i>CFH</i> missense	-	-	-	10	-	7/0,18	–	Tolerated/	0.156	0,26	CFH: 8 KCNT: 5 CFHR3: 5 CFHR1: 1 CFHR4: 1	CFH: 3 CFHR2: 1 CFHR1: 1 F13B: 1

CADD - Combined Annotation Dependent Depletion tool; DNase – DNase hypersensitive regions; Enh – Enhancer; ESE - Exonic Splicing Enhancer, ESS – Exonic Splicing Silencer; Prom – Promoter; Reg – regulatory; TF - Transcript Factor.

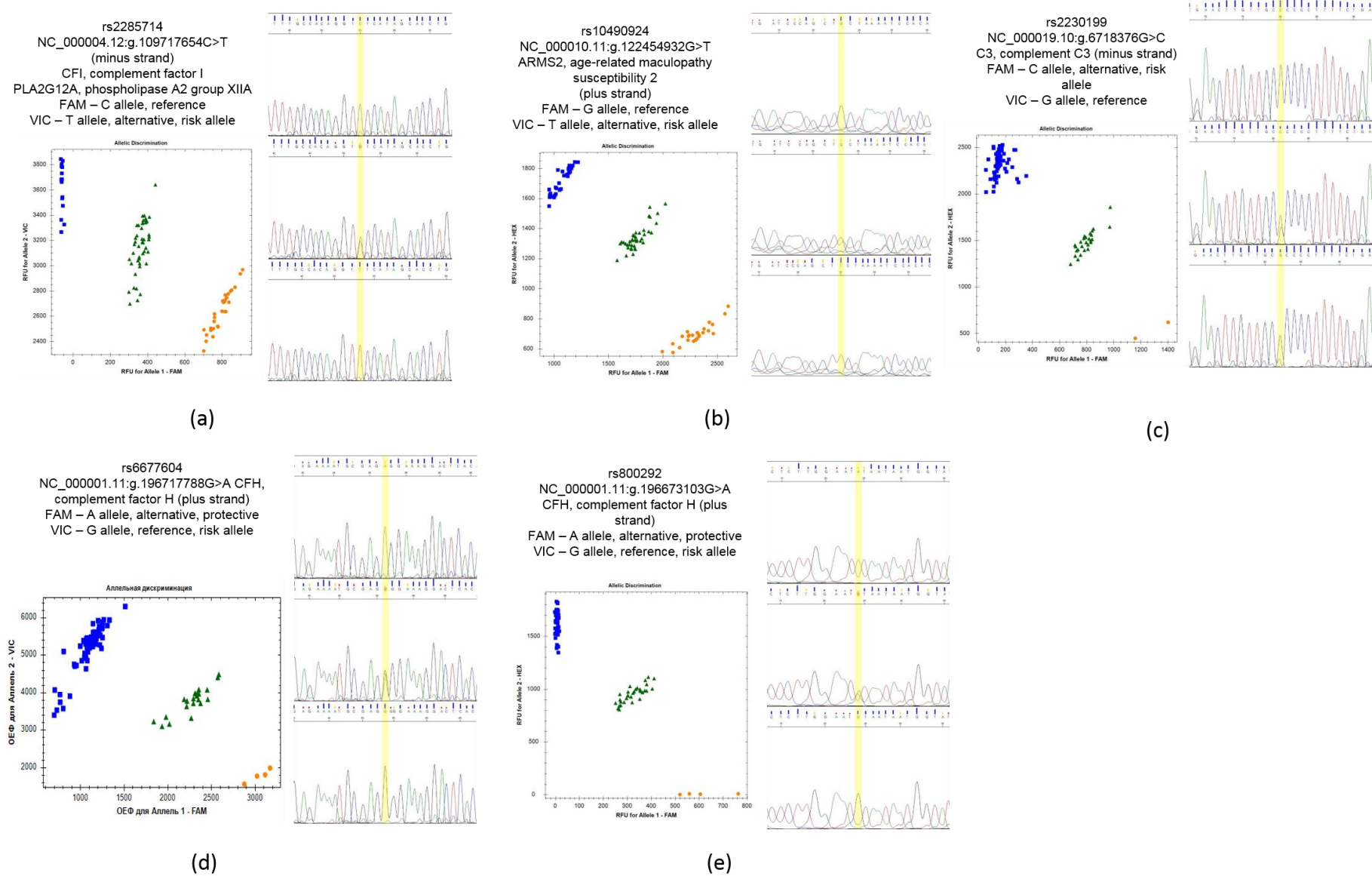


Figure S1. Allelic discrimination and Sanger verification results of rs2285714 (a), rs10490924 (b), rs2230199 (c), rs6677604 (d), rs800292 (e).